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NJHS
National Journal of Health Sciences

Table of Content
Volume 10, Issue 4, 2025

Editorial

220 Transforming Obesity Care: Medications at the Forefront

Tabinda Ashfaq

Research Articles

222 Association of E-Cigarette Use and Oral Health Complications: A Cross Sectional Study in Punjab, Pakistan

Muhammad Taaha, Naila Umer, Hafiz Danish Altaf, Rabia Anjum, Hammad Hassan, Safi Ullah Khan

230 Is Obesity Associated with a Higher Risk of Differentiated Thyroid Cancer in a Cytologically Proven Series of Thyroid Nodules?

Irfan Ahmed Shaikh, Danish Ur Rehman, Sidra Bano, Rajesh Kumar, Ramsha Khalid, Raheem Bakhsh

238 Diagnostic Accuracy of Ultrasound in Detecting Axillary Lymph Nodes in Patients with Early Breast Cancer Taking Histopathology as a Gold Standard

Ambreen Shaikh, Saleha Anwar, Hasnain Ali, Raisa Altaf Malik, Humaira Erum, Bisma Rizwan

244 Role of Heparin to Prevent Hypertrophic Scarring in Donor Area of Split Thickness Skin Graft

Asma Ishtiaq, Faisal Akhlaq Ali Khan, Farhan Zahid, Iqra Muhammad Tasleem, Asra Zahid, Nasreen Bano Khatri

251 Outcomes of Intestinal Anastomosis with or without Platelet Rich Plasma Application

Syeda Wajiha Mustafa, Malik Asad Munir, Atika Tahir, Batool Fatima, Muhammad Azam Mirza, Muhammad Kashif Bashir

258 Risk Assessment of Venous Thromboembolism in Pregnancy and Adequate Prophylaxis: An Observational Study

Aliya Nasim, Sagheera Anjum Manaver, Naima Shah, Humaira Tahir, Mehvish Kanval

264 Experience in Pediatric Bleeding Disorders in Tertiary Care Health Services from Southern Region of Saudi Arabia: Single Center Retrospective Study

Badriah Gharamah Al Asmari, Ali Mujtaba Tahir, Mohammed Alpakra, Mohaned Mustafa Ahmed Mohammad, Sara Saeed Mahamoud Hassanien, Mahnoor Saeed, Manea Abdullah Alqarni, Sami Saleh Alamri

271 Prevalence of Depression and Anxiety in Patients with Obstructive Lung Disease and Correlation with Spirometry

Ehab Nabeel, Sohail Akhtar, Ahmed Ali, Syed Wajahat Hussain, Mujahid Hussain

279 In-Hospital Outcomes of Early Extubation in Post CABG Patients: A Retrospective Study from a Tertiary Care Hospital in Karachi, Pakistan

Qaiser Aziz Khan, Latifa Haidari, Imran Khan Sandeelo, Marium Nastaeen Sarhandi, Sarib Bin Yasir, Mariam Amir

287 Knowledge, Attitudes & Perceptions towards Premenstrual Syndrome among Low Socioeconomic School-Going Adolescent Girls in Karachi, Pakistan

Shaikh Ali Masood, Shabbir Ahmed Mallick, Hamna Jamal, Maria Murtaza, Manahil Nadeem, Ameera Shabbir

292 Timing of Surgical Intervention and its Impact on Short-term Outcomes in Pediatric Hydrocephalus

Shakeel Ahmed, Shoaib Saleem Khan, Syed Zahid Hussain Shah, Muhammad Ali Waqas, Ubaid Ullah, Zeeshan Karim

Case Report

299 Spontaneous Resorption of Mandible in a Middle Aged Female

Hafiz Muhammad Bilal Siddiqui, Hafiz Murtaza Hussain, Talha Ahmed Qureshi, Mariam Mubashir

Research Article

Experience in Pediatric Bleeding Disorders in Tertiary Care Health Services from Southern Region of Saudi Arabia: Single Center Retrospective Study

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Abstract: Background: Bleeding disorders are commonly encountered in clinical practice worldwide. In Saudi Arabia, such disorders are witnessed as a notable entity in clinical practice. They encompass a variable group of disorders – categorized into acquired and congenital. Notable among them are hemophilia, von Willebrand disease (vWD), platelet-function defects, clotting factor deficiencies and immune thrombocytopenia (ITP).

Objective: To describe the various bleeding disorders among Pediatric population and its related clinical features, diagnostics and management modalities adhered to.

Materials and Methods: Retrospective cross-sectional study was carried out in Pediatric Medicine Department, Armed Forces Hospital Southern Region (AFHSR), Khamis Mushait, Kingdom of Saudi Arabia. The data was gathered for last five years – 1st May 2020 to 30th April 2025, as pediatric patients were included who were diagnosed with a bleeding disorder while aged up till fourteen years. Pertinent details subject to relevant history, clinical course and laboratory investigations like blood indices, prothrombin time (PT), activated partial thromboplastin time (PTTK) and accordingly relevant investigations notably factor assay and platelet-function tests were recorded.

Result: During the last five years, 205 patients were evaluated by Pediatric Hematology team in regards to suspicion for a bleeding disorder. Out of these, 21% cases (44 patients) were confirmed with a definite bleeding disorder. Among these 44 patients, 24 (54%) patients being male and 20 (46%) female, all with Saudi ethnicity with their average age of 8 years 4 months. They were diagnosed with a bleeding disorder at average age of 4 years 10 months. 18 (41%) were having an acquired disorder while 26 (59%) were diagnosed with congenital disorder. Chronic ITP was observed in 9 cases while 5 cases were witnessed for each of acute ITP, vWD and factor VII deficiency. The most frequent presenting symptom was petechiae (25% patients) followed by epistaxis in 16% and ecchymosis in 11%, with no significant variation observed in congenital vs acquired disorders. Each patient had an average of one admission during the course of the disease. Thrombocytopenia was the most common laboratory abnormality identified (52% cases). This was followed by abnormal clotting factor levels (27% patients), raised PTTK in 25% and high INR in 14%. Furthermore, WES (whole exome sequencing) was performed in selected cases and genetic mutations were found in 41%. IVIG (intravenous immunoglobulin) was the most common intervention adopted.

Conclusion: We were able to identify a specific bleeding disorder in 21% of cases who were worked up for a suspected bleeding disorder. Among the pediatric population from southern region of Saudi Arabia, chronic ITP was most frequent (20%), followed by 11% cases each for acute ITP, vWD and factor VII deficiency. Awareness strategies aimed towards bleeding disorders accompanied by swift diagnosis, appropriate therapeutics and genetic counseling is indispensable in averting morbidity.

Keywords: Bleeding disorders, Clotting factor, Coagulation profile, Hemophilia, Platelets, von Willebrand disease.

INTRODUCTION

Bleeding disorders are regarded as a global health concern which entail people of divergent ethnicities and ages. Around one in 2,000 adults as well as children in the UK have a diagnosed bleeding disease. Likewise, in Saudi Arabia, such disorders are witnessed as a notable entity in clinical practice. In the Kingdom of Saudi Arabia, no population-based study has examined the prevalence of bleeding disorders. Prevalence in Kingdom will be potentially higher in lieu to much higher rate of consanguinity in Arab ethnicity. These disorders present a complicated challenge in healthcare delivery as well as for research [1-6].

They exhibit a spectrum from familiar inherited conditions as von Willebrand disease (vWD) to acquired entities like immune

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thrombocytopenic purpura, the most common cause of severe thrombocytopenia in pediatric practice. Certain researchers from Saudi Arabia have mentioned the frequency of such medical ailments. Their efforts inferred platelet disorders, von Willebrand disease (vWD) and hemophilia stand amongst the most prevalent bleeding disorders. vWD has a prevalence of 1.5 to 3.9% in Saudi Arabia while 0.08% for Haemophilia A diagnostics and therapeutics adopted in bleeding disorders now follows a definite pathway in developed world. However, the knowledge and facilities for accurate diagnosis and sustained therapy bear various challenges in developing countries [7-12].

Our study, while defining the frequency and clinical aspects of hemostatic disorders, can prove to be pivotal in establishing the effective clinical interventions and instigating constructive public policies. This can steer towards surfacing a holistic approach towards such challenging Pediatric cases in Saudi Arabia. The aim is to define a holistic approach while approaching such disorders in Pediatric population.

MATERIALS AND METHODS

We carried out a retrospective cross-sectional study at Armed Forces Hospital Southern Region (AFHSR), Khamis Mushait, a tertiary care referral center, located in the southern Saudi Arabia. AFHSR serves as a prominent healthcare facility within the Armed Forces Hospital system, catering to military personnel and their dependents residing in the surrounding area. The data was gathered for last five years – 1st May 2020 to 30th April 2025 after permission was sought from hospital ethics committee (REC certificate number AFHSRMREC/2025/DEPARTMENT OF PEDIATRICS/803). Target population was selected by convenience consecutive sampling for pediatric patients up till the age of 14 years, with a diagnosis of bleeding disorder and following with our Pediatric Hematology team.

As our study being a retrospective in design, the limitation to data collection was negotiated with thorough examination of medical archives. Confidentiality was cinched at all tiers. Detailed information from hospital electronic medical system of such children with bleeding disorder was gathered. The hospital possesses robust information and medical data management systems including archives system that aided in data collection needs for this study. A well-structured data collection pro-forma was devised for collecting information on demographic characteristics like age, gender and residential area along with clinical presentations including symptoms, diagnostic modalities utilized – like coagulation profile, platelets count, clotting factors levels. Moreover, treatment details such as interventions administered and hospital inpatient was documented. Confidentiality measures were held fast to ensure patient privacy and data was securely stored for subsequent assessment. The data was analytically evaluated for descriptive statistics, like frequency, mean and standard deviation to outline demographic data, clinical presentations, treatment and outcomes. P value ≤0.05 regarded as significant.

RESULT

During the last five years, 205 patients were evaluated by Pediatric Hematology team in regards to suspicion for a bleeding disorder. Out of these, 21% cases (44 patients) were confirmed with a definite bleeding disorder (representing 0.006% of total 7220 admissions in Pediatric Ward). Among these 44 patients, 24 (54%) patients were male and 20 (46%) female. All were from Saudi ethnicity with their average age of 8 years 4 months. They were diagnosed with a bleeding disorder at average age of 4 years 10 months – with ranging from 1 month to 13 years 2 months. 18 (41%) were having an acquired disorder while 26 (59%) were diagnosed with congenital disorder (as demonstrated in Fig. **1**). Table **1** exhibits the types of bleeding disorders observed in our cases. Chronic ITP was observed in 9 cases while 5 cases were witnessed for each of acute ITP, vWD and factor VII deficiency. 7 patients of congenital disorder were having a family history of bleeding disorders.

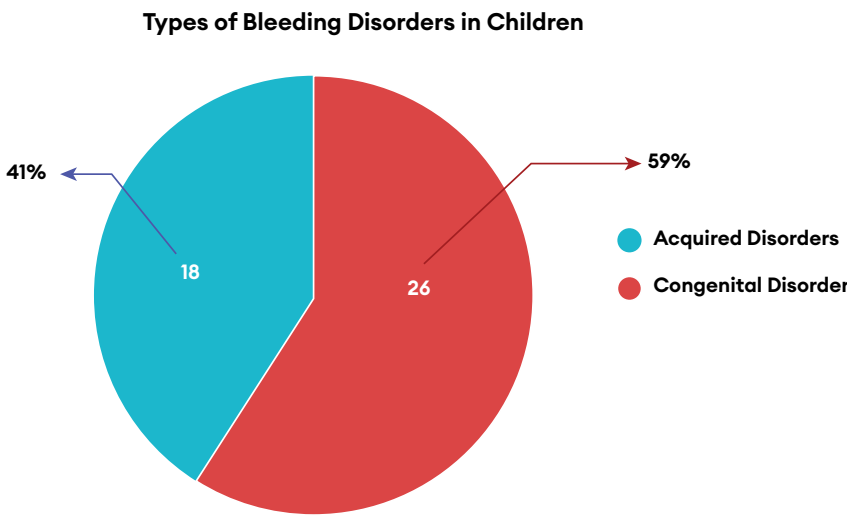

Fig. (1). Types of Bleeding Disorders among our Pediatric Patients (Congenital vs Acquired) (n=44).

Table 1. Type of Bleeding Disorders among our Pediatric Patients (n=44).

Diagnosis	Number of Patients n (%)
Acquired Disorders	
Chronic ITP	9 (20%)
Acute ITP	5 (11%)
Chronic thrombocytopenia unexplained	3 (7%)
Non-Alloimmune thrombocytopenia (NAIT)	1 (2%)
Congenital Disorders	
vWD (von Willebrand disease)	5 (11%)
Factor VII deficiency	5 (11%)
Haemophilia A	4 (9%)
Haemophilia B	1 (2%)
Gray platelet syndrome	1 (2%)
THAMY (Thrombocytopenia, Anemia, and Myelofibrosis) disease	1 (2%)
Congenital amegakaryocytic thrombocytopenia	1 (2%)
Chronic genetic thrombocytopenia	1 (2%)
Bernard-Soulier syndrome	1 (2%)
Fanconi Anaemia	1 (2%)
Afibrogenimia	1 (2%)
Factor XII (Hageman factor) deficiency	1 (2%)
Combined Factor V and VIII deficiency	1 (2%)
Combined Factor VII and X deficiency	1 (2%)
Combined Factor X and XII deficiency	1 (2%)

5 out of 44 patients were noted to have a syndrome (Table **2**). Four cases were having a comorbid disease (two with Type 1 diabetes mellitus and one each for sickle cell disease and G-6-PD deficiency).

Table 2. Type of Syndromes Identified with Bleeding Disorders (n=5).

Syndrome	Bleeding Disorder	Number of Cases n (%)
VATERAL anomaly	Gray platelets syndrome	1 (20%)
Digeorge syndrome	Acute ITP	1 (20%)
Down syndrome	Chronic ITP	1 (20%)
Cornelia de Lange syndrome	Chronic genetic thrombocytopenia	1 (20%)
Arnold Chiari malformation	Chronic ITP	1 (20%)

The presenting symptoms documented in these disorders are depicted in Table **3**, with petechiae being most frequent (25% patients) followed by epistaxis in 16% and ecchymosis in 11%, with no statistical significance among congenital and acquired disorders. Each patient had an average of one admission during the course of the disease with quite variation observed among individual patients (maximum four admissions vs nil admission in certain cases). PICU admission was required once in lieu to intra-cerebral hemorrhage. No other significant complication was observed.

Table 3. Presenting Symptoms in Bleeding Disorders (n=44).

Presenting Symptoms in Bleeding Disorders	Number of Patients n (%)		p-value
	Acquired Disorder	Congenital Disorder	
Petechiae	6 (14%)	5 (11%)	0.238
Istaxis	2 (4%)	5 (11%)	0.388
Skin bruising / ecchymosis	1 (2%)	4 (9%)	0.308
Status post circumcision	0	3 (7%)	0.196
Incidental (pre-operative workup)	0	3 (7%)	0.196
Asymptomatic	1 (2%)	2 (4%)	0.638
Incidental	0	3 (7%)	0.196
Intra-cerebral hemorrhage	0	2 (4%)	0.344
Per rectal bleed	0	2 (4%)	0.344
Gingival bleed	0	2 (4%)	0.344
Hematoma (gluteal)	0	1 (2%)	0.591
Hematuria	0	1 (2%)	0.591
Bleeding per vaginal	0	1 (2%)	0.591

Continue

Continue

Menorrhagia	0	1 (2%)	0.591
Umblical bleed	0	1 (2%)	0.591
Spontaneous conjunctival bleed	0	1 (2%)	0.591

During the laboratory workup in order to strike a definitive diagnosis for bleeding disorder, thrombocytopenia was the most common abnormality identified (52% cases). This was followed by abnormal clotting factor levels (27% patients), raised PTTK in 25% and high INR in 14% (Table **4**). Furthermore, WES (whole exome sequencing) was performed in selected cases and genetic mutations were found in 41%.

Table 4. Abnormal Laboratory Investigations Witnessed in Bleeding Disorders (n=44).

Abnormal Laboratory Investigation	Number of Patients n (%)
Thrombocytopenia (<150 x 109)	23 (52%)
Low clotting factor levels	12 (27%)
Raised PTTK	11 (25%)
High INR (Raised PT)	6 (14%)
Abnormal vWF profile	3 (7%)
Abnormal platelet morphology (Giant platelets)	2 (4%)
Abnormal PFA (Platelet function assay)	1 (2%)
Abnormal WES (Whole exome sequencing)	18 (41%)

The role of investigations like platelets count, PT, PTTK, PFA, clotting factor levels and WES in reaching a final diagnosis is portrayed in Table **5**, which exhibited that basic screening coagulation tools can direct in almost all cases towards a definitive diagnosis.

Table 5. Mandatory Requirement of WES (Whole Exome Sequencing) in Diagnosis of Acquired vs Congenital Bleeding Disorders (n=44).

Type of Bleeding Disorder	Types of Laboratory Investigations required in Confirmation of Diagnosis (n=44)		p-value
	Platelets Count, PT, PTTK, PFA, Clotting Factor Levels	WES (whole Exome Sequencing)	
Acquired	17 (39%)	0 (0%)	0.074
Congenital	22 (50%)	5 (11%)	

Table **6** describes the therapeutic intervention done in various cases of bleeding disorders.

Table 6. Therapeutics Administered in Bleeding Disorders (n=44).

Therapeutic Modality Administered	No. of Patients with Diagnosis				
	Acute ITP	Chronic ITP	NAIT	Hae-mophila A	vWD
IVIG*	2	6	1		
Low dose steroids		3			
Methylpredniso-lone pulse therapy		3	1		
Platelets transfusion	1	1	1		
Sirolimus		1			
Emicizumab				1	
Recombinant Factor VIII				2	
Haemate P					2
Eltrombopag		1			

*Intravenous Immunoglobulins.

DISCUSSION

Bleeding disorders embody a variety of causes as well as discrete symptoms. Certain studies have explored the genetic perspective of such diseases and specific genetic mutations have been implied in coagulation factor defects. The incessant transformation in molecular tools has provided refined diagnostic techniques as well as early detections – inferring in improved therapeutic outcomes. Having a certain diagnosis is crucial for opting right therapeutics as well as for genetic counseling. These disorders have worldwide presence and region of Saudi Arabia stands as no exception [13-18]. Our current study strived to uncover the clinical presentations and involved diagnostics as well as therapeutics employed in our Kingdom specifically in terms of Pediatric bleeding disorders.

While investigating the children with a suspected bleeding disorder, we were able to arrive at a specific bleeding disorder in 21%

cases (44 patients). Among these 44 patients, 24 (54%) patients were male and 20 (46%) female. Yildiz *et al.* while studying bleeding disorders had reported similar observation as 52.5% being male. However, Revel-Vilk *et al.* has published 46% as male gender [19, 20].

Our all cases were from Saudi ethnicity with their average age of 8 years 4 months. They were diagnosed with a bleeding disorder at average age of 4 years 10 months – with ranging from 1 month to 13 years 2 months. Revel-Vilk *et al.* found the median age of 6.4 years at the time of presentation for bleeding disorders [20].

In general, early presentation especially during infancy points towards congenital cause [21]. In our observation, 18 (41%) were having an acquired disorder while 26 (59%) were diagnosed with congenital disorder. Family history can definitely provide a clue in favor of congenital bleeding disorders [22]. 7 (25%) patients of our congenital disorder were having a family history of bleeding disorders. Yildiz *et al.* found family history of bleeding disorder in 19% patients [19]. As the genetic factor is one of the established cause of bleeding diseases, high rate of consanguinity magnifies the prevalence of congenital bleeding disorders among the Saudi people. Awareness by incorporating premarital screening and counseling services can extend comprehensive understanding among the general public [23].

Our research demonstrated chronic ITP in 9 cases while 5 cases were witnessed for each of acute ITP, vWD and factor VII deficiency. While studying the hereditary bleeding disorders, Kannan *et al.* had mentioned hemophilia A to be the commonest congenital hemostatic defect (56% cases) [24]. Yildiz *et al.* found von Willebrand disease to be most frequent (12.1%), followed by hemophilia (9.1%) [19].

Among our studied subjects, petechiae was the most frequent presenting symptoms (25% patients), followed by epistaxis in 16% and ecchymosis in 11%. In contrary, Yildiz *et al.* while documenting the considerable bleeding symptoms, reported epistaxis in 36.4% cases, easy bruising in 32.3% and menorrhagia in 6.1% [19]. Kannan *et al.* reported that the commonest clinical manifestation was subcutaneous hematoma.24 Revel-Vilk *et al.* reported spontaneous bruising as most common symptom (18% patients) [20]. We did not experience any long term complication particularly, anemia, joint deformity and hemiplegia. It is explainable by early diagnosis and the proper handling of specialists in Hematology team.

These diseases are a key mechanism of both morbidity and mortality. It was quite evident that laboratory assessment was a key factor in the diagnosis and management of the disease [25]. 52% cases were spotted to have thrombocytopenia. An elevated level of clotting factor was seen in 27% patients, elevated level of PTTK in 25% and high level of INR in 14%. Yildiz *et al.* discovered that screening tests (platelets, PFA-100, PT, PTTK) were useful in the diagnosis of the bleeding disorder in 36.4 percent patients [19].

STRENGHT AND LIMITATION

Our study has received a validity and strength by the fact that our hospital is a tertiary referral center in the south of Saudi Arabia. Our research is distinguished in regards to its eccentric emphasis on pediatric cases. These findings disclosed valuable insights into factors underlying Pediatric bleeding disorders. This can help in extending efficacious therapeutic interventions and devising the blueprints of public health. The apparent limitation of our study is its retrospective design and relatively smaller number of study participants.

CONCLUSION

We were able to identify a specific bleeding disorder in 21% of cases who were worked up for a suspected bleeding disorder – making 0.006% of total 7220 admissions in Pediatric Ward. Our study surfaced chronic ITP as most frequent (20%), followed by 11% cases each for acute ITP, vWD and factor VII deficiency. This endeavor towards the diverse bleeding disorders in Kingdom of Saudi Arabia provides valued insights into their prevalence, clinical features and adopted therapeutic strategies. Awareness campaigns targeting masses and physicians can serve in prompt diagnosis and timely provision of multiple unanswered queries of families. Further pursuits will definitely aide in achieving the desired goal of advance yet personalized medicine, especially in the emerging framework of carrier detection and gene therapy in varied genetic bleeding disorders.

ABBREVIATIONS

ITP: Immune Thrombocytopenia.

IVIG: Intravenous Immunoglobulin.

PT: Prothrombin Time.

PTTK: Partial Thromboplastin Time.

vWD: von Willebrand Disease.

WES: Whole Exome Sequencing.

AUTHORS' CONTRIBUTION

Badriah Gharamah Al Asmari and **Manea Abdullah Alqarni**: Conceptualization.

Ali Mujtaba Tahir: Writing draft, Critical review and revision the manuscript, Methodology, Data analysis and interpretation.

Mohammed Alpakra and **Sara Saeed Mahamoud Hassanien**: Study design.

Mohaned Mustafa Ahmed Mohammad and **Sami Saleh Alamri**: Methodology, Data analysis and interpretation.

Mahnoor Saeed: Final approval, final proof to be published.

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ETHICAL DECLARATIONS

Data Availability Statement

Data are available upon reasonable request. The data used to support the findings of this study are available from the corresponding author upon request.

Ethical Approval

The study was permitted by the Research Ethics Committee of Armed Forces Hospital Southern Region (AFHSR), Khamis Mushait, Saudi Arabia (REC certificate number AFHSRM-REC/2025/DEPARTMENT OF PEDIATRICS/803).

Consent to Participate

Informed consented.

Consent to Publication

Consented.

Conflict of Interest

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Competing Interest/Funding

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Use of AI-Assisted Technologies

The authors declare that no generative artificial intelligence (AI) or AI-assisted technologies were utilized in the writing of this manuscript, in the creation of images/graphics/tables/captions, or in any other aspect of its preparation.

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